

Health Care Provider Fact Sheet

Disease Name

Isovaleric acidemia

Alternate name(s)
Acronym

Isovaleric acid CoA dehydrogenase deficiency
IVA

Disease Classification

Organic Acid Disorder

Variants

Yes

Variant name
Symptom onset

Chronic intermittent form
Infancy (in the acute neonatal form). The chronic intermittent form presents later in infancy or in childhood.

Symptoms

Episodic overwhelming illness with vomiting, ketosis, acidosis and coma. Hematological abnormalities include leucopenia, thrombocytopenia and possible anemia.

Natural history without treatment

About 50% of patients with the acute neonatal form will die during their first episode. Survivors may have neurological damage though several have made complete recoveries. Patients with the chronic form may have neurologic damage, but the majority of patients are developmentally normal.

Natural history with treatment

Intellectual prognosis depends on early diagnosis and treatment and subsequently on long-term compliance. If treated appropriately, most will have normal development.

Treatment

Low protein diet with restricted leucine intake, glycine supplementation and possible carnitine supplementation.

Emergency Medical Treatment

See sheet from American College of Medical Genetics (attached) or for more information, go to website: http://www.acmg.net/resources/policies/ACT/ACT-sheet_C5_5-3-06.pdf

Other

Sometimes a "sweaty feet" odor is reported during an acute crisis.

Physical phenotype
Inheritance
General population incidence
Ethnic differences
Population
Ethnic incidence

No obvious dysmorphic features.
Autosomal recessive
1:230,000
None known
N/A
N/A

Enzyme location
Enzyme Function

N/A
Isovaleryl-CoA dehydrogenase is the first step in the branched chain organic acid metabolism of leucine.

Missing Enzyme
Metabolite changes

Isovaleryl-CoA dehydrogenase
Urinary isovaleryl glycine, 3-hydroxysoroline acid, increased isovaleric acid in blood. During acute attacks, 4-hydroxyisovaleric acid, mesaconic acid, and methylsuccinic acid, isovalerylglycine and 3-hydroxyisovaleric acid are present. Enzyme analysis by GCMS in amniotic fluid or CVS tissue.

Prenatal testing

Enzyme analysis by GCMS in amniotic fluid or CVS tissue.

MS/MS Profile

Elevated C5 isovaleryl carnitine

OMIM Link
Genetests Link
Support Group

www.ncbi.nlm.nih.gov/entrez/dispomim.cgi?id=243500
www.genetests.org
Organic Acidemia Association
www.oaanews.org
Save Babies through Screening Foundation
www.savebabies.org
Genetic Alliance
www.geneticalliance.org